

## **Haemachromastosis Briefing for Blood Health National Oversight Group**

### Background

In May 2018, policy officials from Welsh Government met with officials from Haemachromatosis UK charity. The purpose of the meeting was to discuss possible under-diagnosis of genetic Haemachromastosis in Wales and subsequent management of the disease.

The agreement from that meeting was for a literature review to be commissioned by Welsh Government to explore in more detail the evidence base surrounding under-diagnosis and gain some clinical perspective on the issues (please see attached).

The opinions of a few Haematology Consultants was sought from South Wales and the general consensus was that in the vast majority of cases, patients are presenting before end-organ damage develops and that late diagnosis does not seem to be a major problem.

In July 2019, there was a debate in the House of Commons regarding genetic Haemachromatosis including the issue of under-diagnosis. It acknowledged the publication of the British Society of Haematology guidelines on management of Haemachromatosis in 2018, that NHSBT encourage people with stable disease to donate blood and recognised the need to raise awareness about GH amongst healthcare professionals.

### Haemachromatosis UK

The charity have made several suggestions regarding work that could be undertaken in Wales. These include:-

- Adherence to recent BSH guidelines
- To ascertain how many people in Wales have a diagnosis of GH and use that to estimate number likely to be under-diagnosed
- Automatic screening of family members (with consent)
- Adding ferritin and transferrin saturation to health check at age 50
- Care package including bilingual literature, ongoing appointments to discuss issues and a central point of contact
- General awareness public campaign including strategy to reach remote communities and those less likely to seek medical attention

With regards to the health check and screening, this is not an issue for the NOG to discuss as this needs to be done at a national level.

### Recommendations

- Assurance that the BSH guidelines are adhered to in Wales

- To gain wider clinical views on whether under-diagnosis is a major issue in Wales
- WBS to encourage people with stable Haemachromatosis to become blood donors
- To monitor for emerging evidence with regards to GH and respond as required